CONTENTS

Preference is given to letters commenting on contributions published recently in the *JRSM*. They should not exceed 300 words and should be typed double spaced.

The genetic background of anticipation

The review article by Dr P Teisberg (April 1995 *JRSM*, pp 185–187) is a timely reminder to clinicians that new concepts in molecular genetics can explain some of the phenomena which have been observed and recorded for decades. Anticipation, the finding of a genetic condition at progressively earlier ages in successive generations usually associated with increasing severity, is one such example.

His comment that the late Lionel Penrose dismissed the clinical data and considered the finding as an observational and ascertainment bias is well recognized[1]. Yet in the early 1960s when I wished to embark on a clinical genetic investigation into one of the common causes of conductive deafness, otosclerosis, Penrose advised, among other things, the search for anticipation. This work confirmed the autosomal dominant inheritance in most families and revealed anticipation in all but three of 45 parent/child relationships[2]. This disease of the otic capsule, of mesodermal origin, unlike other disorders showing anticipation, is not a neuro-degenerative disease. Regrettably, the chromosomal location of this mutation has not, to the best of my knowledge, been investigated.

More recently, a similar study has been conducted into the inheritance of Ménière's disease and a paper read to the Section of Otology of the Royal Society of Medicine (A W Morrison, 'Anticipation in Ménière's disease' 3 February 1995). Most cases are sporadic but some 10% are familial. From these families, some over four generations, the phenomenon of anticipation was very evident in all the 41 families investigated. DNA has been stored from affected and normal individuals. This work is being done in collaboration with Professor R Williamson and Professor J F Mowbray both of St Mary's Hospital Medical School. The search for a trinucleotide repeat is underway. This condition could be classed as neuro-degenerative since the inner ear is of neuro-ectodermal origin.

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Improving the documentation and appropriateness of cardiopulmonary resuscitation decisions

Hignett and colleagues (March 1995 *JRSM*, pp 136–140) are to be congratulated on their attempt to improve documentation of resuscitation decisions using audit. The number of patients in their audit was so small, however, that it will limit the usefulness of their findings. They work in a department of Medicine for the Elderly, but 87% of elderly patients who have cardiopulmonary resuscitation (CPR) have it in other parts of the hospital. The results would be much more powerful if the audit was extended to include all physicians caring for elderly patients.

In the authors' unit 95–98% of patients die without having CPR. Eighty per cent of patients dying in a London teaching hospital were excluded from CPR[1] and, in another study, 31% of elderly medical patients had 'do not resuscitate' (DNR) decisions[2]. Perhaps the authors should ask why so many of their patients do not have resuscitation, rather than concentrating on the small number who might have inappropriate attempts.

Hignett and colleagues suggest that if patients are adamant in their request for resuscitation, despite their doctors' feeling that it is futile, then they should have CPR. We disagree; there is no legal or ethical obligation to offer a treatment that is not effective[3].

The authors may not be aware of our much more extensive audit[4] which improved documentation of CPR decisions in general and elderly medical patients by introducing guidelines for making and recording decisions. Like them, we found that it was impractical for only senior medical staff to make decisions and consultants usually agreed with DNR decisions made by juniors anyway. In other respects we have found that Doyal and Wilsher's guidelines[3] can be adapted for use in general hospitals, although we are aware that they are not yet fully operational in the hospital where they were developed.

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3 Doyal L, Wilsher D. Withholding cardiopulmonary resuscitation: proposals for formal guidelines. *BMJ* 1993;**306**:1593–6

4 Stewart K, Wagg A, Kinirons M. Does audit improve DNR decision making? *J R Coll Physic Lond* 1994;**28**:318–21

Chopin's illnesses

Kuzemko (December 1994 *JRSM*, pp 769–772) provides a fascinating insight into the nature of Frédéric Chopin's chronic respiratory illness. He ultimately comes to

the conclusion that this was most likely due to α-1 antitrypsin deficiency. He dismisses the suggestion which seems to have been initially made by O'Shea[1] that he and his sibling had cystic fibrosis. The major reason for this is the suggestion that cystic fibrosis was uniformly fatal in the pre-antibiotic era during the first 10 years of life.

Having discussed this with Dr O'Shea, I would urge Dr Kuzemko to reconsider the possibility that Chopin and his sister Emilia both had cystic fibrosis. This diagnosis fits much better with the known facts. Both had lower respiratory illness in the early years of life which is very uncommon in α-1 antitrypsin deficiency. Frédéric eventually developed bronchiectasis which is characteristic of cystic fibrosis whereas in α-1 antitrypsin deficiency the problem is emphysema not bronchiectasis. Haemoptysis must be very uncommon in that condition.

Patients with cystic fibrosis certainly reached adult life in the pre-antibiotic era. For many years the oldest known patient was a man born in Melbourne in 1913 and first reported in 1960 by Marks and Anderson[2]. For the first 20 years of his life there were no sulphanomides and for the first 30 no antibiotics. He eventually died at the age of 66 from disseminated carcinoma of the colon. There is a great variability in the pulmonary manifestations of cystic fibrosis. Supporting the diagnosis of cystic fibrosis in Frédéric was the suggestion that he had malabsorption. At the age of 28 he was well below the third centile, again uncommon in α-1 antitrypsin deficiency which usually does not produce symptoms until the fourth or fifth decade. He seems to have had delayed puberty and despite evidence of a number of sexual partners, there is no record of his having fathered a child. Again, this is consistent with the infertility so characteristic of the male with cystic fibrosis.

It is well recognized that some patients with cystic fibrosis can have chronic liver disease with minimal respiratory problems. Emilia's recurrent and eventually fatal haematemesis could well have been bleeding from oesophageal varices secondary to the chronic liver disease of cystic fibrosis.

After 150 years it is impossible to be certain as to the nature of the illness of Frédéric Chopin but speculation on the health problems of the famous remains a fascinating intellectual exercise. I believe the evidence strongly supports the suggestion of O'Shea that cystic fibrosis was the probable cause of his chronic ill health and premature death and also that of his sister.

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Medical ethics

I work for the National Council for Hospice and Specialist Palliative Care Services, and write in response to the letter from Dr Michael H K Irwin (April 1995 *JRSM*, pp 240).

In their evidence to the House of Lords Select Committee on Medical Ethics, Council made it clear that voluntary euthanasia has no place in specialist palliative care. Palliative care focuses on the quality of life, relief of pain and other symptoms, and meeting psycho-social needs. It is estimated[1] that 28 000 people (most with cancer), died in a hospice in 1994, and that well over half of the 160 000 people who die at home are cared for by palliative care services. It is estimated that 95% of pain can be alleviated and pain in the remaining number of patients can be reduced.

This provision and these achievements are usually ignored by those who argue for voluntary euthanasia; Dr Irwin did not mention in his letter his active involvement with the Voluntary Euthanasia Society.

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